The Changing Moral Focus of Newborn Screening

An Ethical Analysis by the President's Council on Bioethics – December 2008

ACHDNC Response

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COB Members by Discipline

- 1 Economics
- 5 Ethics
- 3 Law
- 8 Medicine
- 1 Philosophy
- 2 Political Science
- 1 Psychology

MDs by Specialty

- Endocrinology
- Bioethics
- Genetics
- Nephrology
- Neurology
- Neuroscience research
- Neurosurgery
- Psychiatry

Purpose of the White Paper

- "to foster public awareness of the practice of newborn screening, the ethical principles that have guided it until now and the ethical problems posed by its current and future expansion."

Question and Conclusions

- ***Question***
- "What ethical principles should guide the practice of newborn screening in the United States?"

- ***Conclusions***
- Seven elements were discussed that should be part of "an ethically sound approach to public policy in newborn screening".

Element #1

- *Reaffirm the essential validity and continuing relevance of the classical Wilson-Jungner screening criteria [WHO - 1968]*
  - *The 10 W-J criteria for population-based screening are summarized by "screen only if you can treat"*
  - *ONLY the W-J criteria should guide NBS*
- Implications
  - The core panel may not meet the W-J criteria
  - Evidence-based decision-making lacking
  - Additions to the core panel may not meet criteria
  - Other criteria has no bearing on NBS

Forty Years of Progress

- 1975 “Genetic Screening” Report
  - National Research Council of the National Academy of Sciences [NAS/NRC]
  - Broadened the concept of “BENEFIT” in NBS:
    - 1. Direct medical treatment to the infant
    - 2. To facilitate management decisions
    - 3. Provide supportive treatment to the infant
    - 4. Inform subsequent reproductive decisions
    - 5. Provide knowledge regarding rare diseases

10 Years of Progress

- 1990s Tandem mass spectrometry
- 1991 American College of Medical Genetics
- 2003 Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children
- 2005 HRSA/ACMG Core Panel
- 2006-8 ACHDNC Workgroup Reports
  - Nomination Review and Prioritization WG
  - Internal Review WG
  - External Evidence Review WG
  - Decision Criteria & Process WG

ACMG Expert Group – Core Panel

- NBS policy should be driven by "what is best for the affected infant".
- Both Wilson-Jungner and NAS/NRC criteria used:
  - Specific and sensitive screening test
  - Sufficiently well understood natural history
  - Available and efficacious treatment
    - Infant: management and support
    - Family: inform subsequent reproductive decisions
    - Society: Knowledge about condition [dec "odyssey"]
  - A benefit to RESEARCH studies was NOT a criteria
- STATES will make the final decisions

Decision & Criteria WG Report

- 2009 – Method for evaluating conditions nominated for population-based screening of newborns and children
  - Addressed current and future unique issues in NBS
    - Multiplex technology
    - Development of innovation and new information
    - “Benefit” and assessment of evidence in conditions with limited population-based controlled trials

Element #1 Response

- The ACMG criteria for inclusion to the currently recommended panel of core conditions are consistent with the Wilson-Jungner and NAS/NRC principles.
  - There is documented benefit to the affected infant from early detection
  - There is a reliable screening test that is feasible in a public health setting

Element #2 / Response

- *Insist that mandatory NBS be recommended to states only for those disorders that clearly meet classical criteria.*

- The 29 core conditions do meet appropriate criteria [W-J and NAS/NRC].

- "Secondary" conditions are laboratory findings incidental to the testing procedure or as a consequence of clarifying the differential diagnosis of a core condition.

Element #3

- *Endorse the view that screening for other conditions that fail to meet classical criteria may be offered by the states to parents on a voluntary basis under a research paradigm.*

- *"Classical criteria" is limited to the original 10 Wilson-Jungner criteria*
- *Cited the Massachusetts experience*
  - *10 core mandatory conditions*
  - *All other conditions optional*

Element #3 Response

- There is a need to move forward with appropriate application of the Wilson-Jungner, NAS/NRC, and ACHDNC criteria.

- When conditions to not meet those expanded criteria, there is clearly a role for research within NBS programs to enhance screening techniques and study disorders that may be candidates to join the recommended core panel.

Element #4

- *Affirm that when the differential diagnosis of some targeted disorders entails detection of other poorly understood conditions [that would not otherwise be suitable candidates for NBS, such results need NOT be transmitted to the child's physician or parents.*

- *Individual states may choose to:*
  - *Suppress the information*
  - *Obtain informed consent at the time of the NBS*

Element #4 – Response

- These are truly incidental and inevitable findings that are an integral part of the testing process for the core panel

- Why reveal incidental findings?
  - It is unfair/unreasonable to disregard these results
  - Avoid the "diagnostic odyssey"
  - Inform reproductive decision-making
  - Early supportive intervention for the child and family
  - Clinical research studies may be available to family

- Informed consent
  - Not appropriate for the core conditions
  - Required for research studies
  - Confusing for incidental findings – risk for NBS

Element #5 / Response

- *Encourage the states to reach a consensus on a uniform panel of conditions clearly meriting mandatory screening.*

- The Secretary's Advisory Committee on Heritable Disorders in Newborns and Children is here for you.

Element #6

- *Urge a thorough and continuing re-evaluation of the disorders now recommended for inclusion in the mandatory screening panel, to ascertain whether they genuinely meet the classical criteria that would justify mandatory screening of all newborns, or whether they instead are suitable candidates for pilot screening studies.*

Element #6 – Response

- Continual evaluation of the national newborn screening program is appropriate and ongoing:
  - National Coordinating Center for the Genetics and Newborn Screening Regional Collaborative Groups
  - Newborn Screening Saves Lives Act
  - National Newborn Screening Clearinghouse
  - Advisory Committee on Heritable Disorders in Newborns and Children
  - Advisory Committee on Genetics Health and Society

Element #7 / Response

- *Reject any simple application of the "technological imperative", i.e., the view that screening for a disorder is justified by the mere fact that it is detectable via multiplex assay*

- If all other criteria are met, the review process looks at technology to answer 3 questions:
  - Is a suitable test available?
  - Can that test meet public health needs [national]?
  - Is the test economically feasible?

Conclusion

- NBS is a state-based established and effective public health program – a model for early diagnosis and treatment. The ACHDNC offers guidance through its recommendations to the Secretary.

- The ACHDNC has moved well beyond the seven elements noted in the COB report. The Committee has created a system of structured, evidence-based assessment that supports a consistently rigorous, iterative, and transparent approach to making recommendations regarding broad population-based screening programs for rare conditions in infants and children.